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Glanzmann's Thrombasthenia: Report of A Case and Review of the Literature

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ARTICLE DETAILS

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ABSTRACT

Glanzmann's thrombasthenia (GT)is a rare autosomal recessive disorder characterized by qualitative or quantitative abnormalities of the platelet membrane glycoprotein (GP) IIb/IIIa. These integrins are encoded by the ITGA2B and ITGB3 genes and form platelet glycoprotein (GP)IIb/IIIa, which acts as the principal platelet receptor for fibrinogen. Spontaneous mucocutaneous bleeding is common and can lead to fatal bleeding episodes.

1. Introduction

Platelets are a central component of many restorative physiological processes, including hemostasis. During hemostasis, damaged sub endothelium releases adhesive proteins (ie, collagen and thromboplastin) and fibrinogen, which bind with aggregated platelets at the site of injury, forming a platelet plug. Platelets then provide a surface and phospholipid source for attachment of coagulation cofactors. Subsequent activation of the coagulation pathways prompts fibrin attachment to activated platelets, creating a thrombus. Any disruption in platelet function, whether acquired or inherited, will generate bleeding.

Glanzmann's thrombasthenia (GT) was first documented in 1918 by Dr. Eduard Glanzmann, who described a novel platelet abnormality with defective clot retraction and abnormal appearance on stained film, as "hereditary haemorrhagic thrombasthenia" this syndrome is characterized by mucocutaneous bleeding with a variable clinical presentation ranging from mild bruising to severe and potentially fatal haemorrhages. [[1,5-6,8]. Glanzmann thrombasthenia (GT) is a rare autosomal recessive disorder characterized by a deficiency or functional defect of platelet glycoprotein (GP) IIb/IIIa, which mediates aggregation of activated platelets by binding the adhesive proteins, fibrinogen, Von Willebrand factor (VWF) and fibronectin.[2] GT is rare, with an incidence of approximately 1:1 million, although this is much higher in areas where marriage between close family relatives is common [3,6] United States as those affecting less than 200,000 individuals. The exact incidence has been difficult to calculate, but is estimated at one in 1,000,000. With an autosomal recessive inheritance, males and females are affected equally. [4,6-7]. The common clinical manifestations are epistaxis, gum bleeding, and menorrhagia.[7].

Despite its rarity, it has gained attention since the discovery of its pathophysiology, due to the consequent development of antiplatelet agents now commonly used during percutaneous coronary interventions in this report, we describe a case of Glanzmann's thrombasthenia and review the current literature. Finally, I would like to tell the patient after a treatment his general condition is better, and also, he accepted his diagnosis and he is ready to receive the treatment until his lifetime.

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2. Patient Description

My patient's name is Mr. Anto Ronal, he is 22-year-old, he is a student, As well as he is working part time. his father and mother were a Thalassemia. He is having 12 brother and sisters. 8 males and 4 females, he is the 6th child in his family. In his family except father and mother 4 children they had diagnosis of Thalassemia. But no one is not having these problems of GT. No family history of these diseases.

3. Case History

The patient was 22-year-old male who presented with fever, shortness of breath and fatigue. He had a long clinical history of easy and spontaneous bruising, excessive bleeding with tooth extractions, severe epistaxis as a child, when he was 13-year-old. There was no family history of easy bruising or excessive bleeding. During hospitalization the patient became hypotensive but responded successfully to treatment. However, the epistaxis was persistent and no origin was found.

4. Physical Examination

The patient with GT present with signs of purpura and bleeding. The initial physical examination focused on assessing hemodynamic stability. When he was severe bleeding from the nose hemodynamically unstable. Apart from that the physical examination findings are limited use only.

5. Result of Pathological Test & other investigation

The disorder is caused by deficiency or abnormality of the platelet glycoprotein IIb and/or IIIa. For my patient a normal platelet count on a routine blood smear does not rule out a diagnosis of GT, as patients with GT usually show no abnormalities in the platelet count. The prothrombin time and activated partial thromboplastin time will also be normal. However, the bleeding time will be prolonged, (12 minutes) which warrants further investigation.

Light transmission aggregometry (LTA) is widely accepted as the gold standard diagnostic tool for assessing platelet function (<10%). Platelet Function Analyzer (PFA) is a highly sensitive test for detecting GT. The PFA assay uses collagen + ADP- and collagen + epinephrine-embedded cartridges to mimic a damaged vessel endothelium. As citrated whole blood flows at a high shear stress rate through these cartridges, platelets bind, creating a platelet plug. The PFA assay is prolonged among patients with GT. For my patient it was 100. Flow cytometry can be beneficial, as GT includes glycoprotein receptor deficiency and/or dysfunction. It was (<20%) GPIIb and GPIIIa. Apart from this investigation the doctor was tend to do DNA study also.

Overall, the diagnosis of GT includes the presence of normal platelet count (typically on the lower end of normal), prolonged bleeding time, and prolonged PFA time. Platelets fail to aggregate under the conditions utilized in LTA, which is uniquely indicative of GT.

6. Treatment Plan

The treatment plan for Glanzmann thrombasthenia aims to manage and prevent bleeding episodes, improve platelet function, and maintain overall health. Platelet transfusions are the standard treatment for bleeds in GT that remain refractory to local measures and/or antifibrinolytic drugs, but this treatment may result in the development of antibodies to glycoprotein IIb–IIIa and/or HLA, rendering further transfusions ineffective.

But my patient every month used to receive platelet transfusion and 3 months once he receives Red blood Cell transfusion, and also, he was receiving Injection Novaseven and tablet Tranexamic Acid when he was having bleeding.

Apart from this he needs supportive care, regular check-up, genetic counseling, psychological support, should avoid certain medication like NSAID and anticoagulant, and teaching about the emergency plan for unexpected bleeding.

On the other hand, he can complement the standard treatment plan for Gene therapy, Bone marrow Transplantation, Novel Therapies (such as small molecule inhibitors or monoclonal antibodies targeting specific pathways involved in platelet function and clot formation), and Platelet-targeted Therapies, etc…

7. Expected outcome of the treatment plan

The treatment plan for Glanzmann thrombasthenia, a rare inherited bleeding disorder, typically aims to manage and prevent bleeding episodes, with proper management and adherence to the treatment plan, individuals with Glanzmann thrombasthenia can lead relatively normal lives with reduced risk of bleeding complications. However, the severity of the condition can vary, and the effectiveness of treatment may differ from person to person. Regular communication with healthcare providers is essential for optimal outcomes.

8. Actual Outcome

The actual outcome of Glanzmann thrombasthenia varies from person to person and depends on several factors, including the severity of the condition, the effectiveness of treatment, and individual response to therapy. Overall, while Glanzmann thrombasthenia poses challenges for affected individuals, advancements in medical care and supportive therapies have improved outcomes and quality of life for many patients. However, it's important for individuals with Glanzmann thrombasthenia to work closely with healthcare providers to develop a personalized treatment. plan and receive ongoing support and management**.**

9. References

1. Christopher Sebastiano, Michael Bromberg, Karen Breen, Matthew T. Hurford, "Glanzmann's thrombasthenia: report of a case and review of the literature". Int J Clin Exp Pathol 2010;3(4):443-447
2. Massimo F., Emmanuel F., Giuseppe L. "Glanzmann thrombasthenia clinical management" Clinica Chimica Acta", Volume 4, Issues 1-2, 2010, Elsevier, Page 1-6
3. Man-Chiu Poon,1* Roseline d'Oiron,2* Rainer B. Zotz,3* Niels Bindslev,4* Matteo Nicola Dario Di Minno,5,6* and Giovanni Di Minno5* The international, prospective Glanzmann Thrombasthenia Registry: treatment and outcomes in surgical intervention haematologica | 2015; 100(8
4. Juliana Perez Botero,1 Kristy Lee,2 Brian R Branchford,3 Paul F Bray,4 Kathleen Freson,5 Michele P. Lambert,6 Minjie Luo,7 Shruthi Mohan,2 Justyne E. Ross,2 Wolfgang Bergmeier8 and Jorge Di Paola9 Haematologica 2020, Volume 105(4):888-894.
5. Tia Solh, Ashley Botsford & Melhem Solh, Glanzmann's thrombasthenia: pathogenesis, diagnosis, and current and emerging treatment options, Journal of Blood Medicine, , 219-227, DOI: 10.2147/JBM.S71319.
6. Natalie Mathews1 Georges-Etienne Rivard2 Arnaud Bonnefoy, Glanzmann Thrombasthenia: Perspectives from Clinical Practice on Accurate Diagnosis and Optimal Treatment Strategies, Journal of Blood Medicine, , 449-463, DOI: 10.2147/JBM.S271744.
7. Irem Iqbal1, Saima Farhan2 and Nisar Ahmed2, Glanzmann Thrombasthenia: A Clinicopathological Profile, Journal of the College of Physicians and Surgeons Pakistan 2016, Vol. 26 (8): 647-650.
8. Man C.P, Giovanni D.M, Roseline d O, Rainer Z., New insights into the treatment of Glanzmann Thrombasthenia, Elsevier, transfusion Medicine Review 30(2016) 92-99.